



CYP4V2 gene

cytochrome P450 family 4 subfamily V member 2

Normal Function

The *CYP4V2* gene provides instructions for making a member of the cytochrome P450 family of enzymes. These enzymes are involved in the formation and breakdown of various molecules and chemicals within cells. The CYP4V2 enzyme is involved in a multi-step process called fatty acid oxidation in which fats are broken down and converted into energy, but the enzyme's specific function is not well understood.

Health Conditions Related to Genetic Changes

Bietti crystalline dystrophy

At least 42 *CYP4V2* gene mutations have been identified in people with Bietti crystalline dystrophy, a disorder in which numerous small, yellow or white crystal-like deposits of fatty (lipid) compounds accumulate in the light-sensitive tissue that lines the back of the eye (the retina). The deposits damage the retina, resulting in progressive vision loss.

CYP4V2 gene mutations that cause Bietti crystalline dystrophy are predicted to change the structure of the CYP4V2 enzyme in a way that reduces or eliminates its activity. The mutations likely affect lipid breakdown; however, it is unknown how they lead to the specific signs and symptoms of Bietti crystalline dystrophy. For unknown reasons, the severity of the signs and symptoms differs significantly among individuals with the same *CYP4V2* gene mutation.

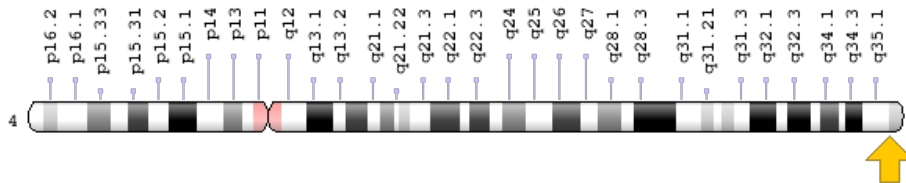
other disorders

Certain common variations (polymorphisms) in the *CYP4V2* gene have been associated with an increased risk of a type of blood clot called a deep venous thrombosis (DVT). DVTs occur most often in the deep veins of the legs or arms. If these clots travel through the bloodstream, they can lodge in the lungs and cause a life-threatening complication called a pulmonary embolism. While nearby genes on chromosome 4 are known to be involved in blood clotting, it is unclear how the *CYP4V2* gene variations may affect this process.

Chromosomal Location

Cytogenetic Location: 4q35.1-q35.2, which is the long (q) arm of chromosome 4 between positions 35.1 and 35.2

Molecular Location: base pairs 186,191,520 to 186,213,463 on chromosome 4 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- BCD
- CP4V2_HUMAN
- CYP4AH1
- cytochrome P450 4V2
- cytochrome P450, family 4, subfamily V, polypeptide 2

Additional Information & Resources

GeneReviews

- Bietti Crystalline Dystrophy
<https://www.ncbi.nlm.nih.gov/books/NBK91457>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28CYP4V2%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D>

OMIM

- CYTOCHROME P450, FAMILY 4, SUBFAMILY V, POLYPEPTIDE 2
<http://omim.org/entry/608614>

Research Resources

- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=CYP4V2%5Bgene%5D>
- HGNC Gene Family: Cytochrome P450 family 4
<http://www.genenames.org/cgi-bin/genefamilies/set/1003>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=23198
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/285440>
- UniProt
<http://www.uniprot.org/uniprot/Q6ZWL3>

Sources for This Summary

- Bezemer ID, Bare LA, Doggen CJ, Arellano AR, Tong C, Rowland CM, Catanese J, Young BA, Reitsma PH, Devlin JJ, Rosendaal FR. Gene variants associated with deep vein thrombosis. *JAMA*. 2008 Mar 19;299(11):1306-14. doi: 10.1001/jama.299.11.1306.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/18349091>
- OMIM: CYTOCHROME P450, FAMILY 4, SUBFAMILY V, POLYPEPTIDE 2
<http://omim.org/entry/608614>
- Li A, Jiao X, Munier FL, Schorderet DF, Yao W, Iwata F, Hayakawa M, Kanai A, Shy Chen M, Alan Lewis R, Heckenlively J, Weleber RG, Traboulsi EI, Zhang Q, Xiao X, Kaiser-Kupfer M, Sergeev YV, Hejtmancik JF. Bietti crystalline corneoretinal dystrophy is caused by mutations in the novel gene CYP4V2. *Am J Hum Genet*. 2004 May;74(5):817-26. Epub 2004 Mar 23.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/15042513>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1181977/>
- Mamatha G, Umashankar V, Kasinathan N, Krishnan T, Sathyabaarathi R, Karthiyayini T, Amali J, Rao C, Madhavan J. Molecular screening of the CYP4V2 gene in Bietti crystalline dystrophy that is associated with choroidal neovascularization. *Mol Vis*. 2011;17:1970-7. Epub 2011 Jul 20.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/21850171>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3154135/>
- Nakano M, Kelly EJ, Rettie AE. Expression and characterization of CYP4V2 as a fatty acid omega-hydroxylase. *Drug Metab Dispos*. 2009 Nov;37(11):2119-22. doi: 10.1124/dmd.109.028530. Epub 2009 Aug 6.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/19661213>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2774980/>
- Nakano M, Kelly EJ, Wiek C, Hanenberg H, Rettie AE. CYP4V2 in Bietti's crystalline dystrophy: ocular localization, metabolism of #-3-polyunsaturated fatty acids, and functional deficit of the p.H331P variant. *Mol Pharmacol*. 2012 Oct;82(4):679-86. Epub 2012 Jul 6.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/22772592>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3463217/>

- Rossi S, Testa F, Li A, Iorio VD, Zhang J, Gesualdo C, Corte MD, Chan CC, Fielding Hejtmancik J, Simonelli F. An atypical form of Bietti crystalline dystrophy. *Ophthalmic Genet.* 2011 Jun;32(2): 118-21. doi: 10.3109/13816810.2011.559653. Epub 2011 Mar 8.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/21385027>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3155699/>
 - Xiao X, Mai G, Li S, Guo X, Zhang Q. Identification of CYP4V2 mutation in 21 families and overview of mutation spectrum in Bietti crystalline corneoretinal dystrophy. *Biochem Biophys Res Commun.* 2011 Jun 3;409(2):181-6. doi: 10.1016/j.bbrc.2011.04.112. Epub 2011 May 1.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/21565171>
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